



ALG1 gene

ALG1, chitobiosyldiphosphodolichol beta-mannosyltransferase

Normal Function

The *ALG1* gene provides instructions for making an enzyme that is involved in a process called glycosylation. During this process, complex chains of sugar molecules (oligosaccharides) are attached to proteins and fats (lipids). Glycosylation modifies proteins so they can fully perform their functions and modifies lipids so they can help cells interact with each other. Oligosaccharides are made up of many sugar molecules that are attached to one another in a stepwise process, forming a complex chain. The enzyme produced from the *ALG1* gene transfers a simple sugar called mannose to growing oligosaccharides at a particular step in the formation of the chain. Once the correct number of sugar molecules are linked together, the oligosaccharide is attached to a protein or lipid.

Health Conditions Related to Genetic Changes

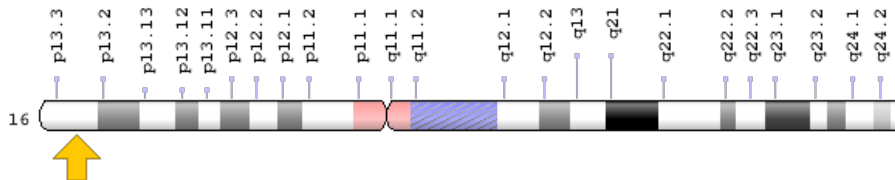
ALG1-congenital disorder of glycosylation

At least 15 mutations in the *ALG1* gene have been found to cause *ALG1*-congenital disorder of glycosylation (*ALG1*-CDG). This condition typically leads to intellectual disability, delayed development, weak muscle tone (hypotonia), and other signs and symptoms that affect many body systems. Mutations in the *ALG1* gene result in the production of an abnormal enzyme with little activity. The poorly functioning enzyme cannot add mannose to sugar chains efficiently, and the resulting oligosaccharides are often incomplete. Although the short oligosaccharides can be transferred to proteins and lipids, the process is not as efficient as with the full-length oligosaccharide. The wide variety of signs and symptoms in *ALG1*-CDG are likely due to impaired glycosylation of proteins and lipids that are needed for normal function of many organs and tissues.

Chromosomal Location

Cytogenetic Location: 16p13.3, which is the short (p) arm of chromosome 16 at position 13.3

Molecular Location: base pairs 5,071,809 to 5,087,379 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- asparagine-linked glycosylation 1 homolog (yeast, beta-1,4-mannosyltransferase)
- asparagine-linked glycosylation 1, beta-1,4-mannosyltransferase homolog
- asparagine-linked glycosylation protein 1 homolog
- beta-1,4 mannosyltransferase
- beta-1,4-mannosyltransferase
- chitobiosyldiphosphodolichol beta-mannosyltransferase
- GDP-Man:GlcNAc2-PP-dolichol mannosyltransferase
- GDP-mannose-dolichol diphosphochitobiose mannosyltransferase
- hMat-1
- HMAT1
- HMT-1
- HMT1
- mannosyltransferase-1
- Mat-1
- MT-1

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (second edition, 2009): Cellular Organization of Glycosylation
<https://www.ncbi.nlm.nih.gov/books/NBK1926/>
- Molecular Biology of the Cell (fourth edition, 2002): Most Proteins Synthesized in the Rough ER Are Glycosylated by the Addition of a Common N-linked Oligosaccharide
<https://www.ncbi.nlm.nih.gov/books/NBK26841/#A2230>
- Molecular Biology of the Cell (fourth edition, 2002): What Is the Purpose of Glycosylation?
<https://www.ncbi.nlm.nih.gov/books/NBK26941/#A2354>
- Molecular Cell Biology (fourth edition, 2000): A Common Preformed N-Linked Oligosaccharide Is Added to Many Proteins in the Rough ER
<https://www.ncbi.nlm.nih.gov/books/NBK21744/#A4826>

GeneReviews

- Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1332>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALG1%5BTIAB%5D%29+OR+%28%28GlcNAc2-PP-dolichol+mannosyltransferase%5BTIAB%5D%29+OR+%28beta-1,4+mannosyltransferase%5BTIAB%5D%29+OR+%28beta-1,4-mannosyltransferase%5BTIAB%5D%29+OR+%28mannosyltransferase-1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ALG1, YEAST, HOMOLOG OF
<http://omim.org/entry/605907>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ALG1%5Bgene%5D>
- HGNC Gene Family: Glycosyl transferases group 1 domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/427>

- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18294
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/56052>
- UniProt
<http://www.uniprot.org/uniprot/Q9BT22>

Sources for This Summary

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